GUSB gene

glucuronidase beta

Normal Function

The *GUSB* gene provides instructions for producing an enzyme called beta-glucuronidase (β-glucuronidase). This enzyme is located in lysosomes, compartments within cells that digest and recycle different types of molecules. β-glucuronidase is involved in the breakdown of large molecules called glycosaminoglycans (GAGs). GAGs are composed of sugar molecules that are linked together to form a long string. To break down these large molecules, individual sugars are removed one at a time from one end of the molecule. β-glucuronidase is involved in the break down of three types of GAGs: dermatan sulfate, heparan sulfate, and chondroitin sulfate. This enzyme removes a sugar called glucuronic acid when it is at the end of the GAG chain.

Health Conditions Related to Genetic Changes

mucopolysaccharidosis type VII

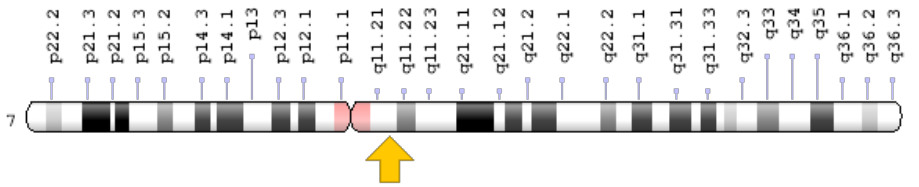
At least 55 mutations in the *GUSB* gene have been found to cause mucopolysaccharidosis type VII (MPS VII). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS VII reduce or eliminate the function of β-glucuronidase.

The shortage (deficiency) of β-glucuronidase leads to the accumulation of dermatan sulfate, heparan sulfate, and chondroitin sulfate within lysosomes in virtually all tissues and organs. The buildup of these GAGs increases the size of the lysosomes, which is why many tissues and organs are enlarged in MPS VII. Researchers believe that the accumulated GAGs may also interfere with the functions of other proteins inside the lysosomes and disrupt many normal functions of cells.

Chromosomal Location

Cytogenetic Location: 7q11.21, which is the long (q) arm of chromosome 7 at position 11.21

Molecular Location: base pairs 65,960,684 to 65,982,314 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- beta-D-glucuronidase
- beta-G1
- beta-glucuronidase
- beta-glucuronidase precursor
- BG
- BGLR_HUMAN
- FLJ39445
- glucuronidase, beta
- MPS7

Additional Information & Resources

Educational Resources

- Eurekah Bioscience Collection: Defects in Glycosaminoglycan Degradation (Mucopolysaccharidoses) https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53462

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GUSB%5BTI%5D%29+OR+%28beta+glucuronidase%5BTI%5D%29%29+OR+%28MPS+VII%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- BETA-GLUCURONIDASE
  http://omim.org/entry/611499

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GUSB.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GUSB%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4696
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2990
- UniProt
  http://www.uniprot.org/uniprot/P08236

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